

The helix and health: applying genetics to primary care

Gene Scene, a new section of *wjm*, aims to help clinicians put genetic principles into practice

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Over the last decade, the development of powerful tools for discovering genes has culminated in the recent publication of the human genome sequence. But with this focus on gene discovery and the prospect of developing new drugs or gene therapies has come a tendency to overlook the practical application of genetics to patient care. Clinicians have at their fingertips all the tools—their clinical skills, the family history, and genetic testing—to identify patients who are at risk for, or who have, a genetic disorder. Yet, physicians are often unfamiliar with basic genetic concepts, and many find genetics baffling. To help illuminate the practical uses of genetics, *wjm* begins *Gene Scene*, a series of articles that will focus on the use of genetic testing in patient care.

We start this series with a discussion of the uses of genetic testing in medical practice and in patient decision making (see p 344). Because patient education and genetic counseling are integral to genetic testing, the second article in the series will explain the process of genetic consultation and the complementary role of genetics professionals and primary care physicians in the care of patients with inherited disorders. The series will continue to its end with examples of the role of genetic counseling and testing in certain common diseases with a heritable component (breast cancer, colon cancer, hearing loss, and Alzheimer's disease) and specific common Mendelian disorders (hereditary hemochromatosis and cystic fibrosis).

When it comes to the "new genetics," clinicians face 2 major sources of confusion as they try to keep up with what is happening. The first involves the language that is used to talk about genes. For example, genetic testing means the analysis of 4 possible types of material—RNA, DNA, chromosomes, or proteins—to understand the basis of an inherited disorder. In our current lexicon, however, molecular genetic testing (the analysis of DNA or RNA) has become virtually synonymous with the term *genetic testing*. This is unfortunate because it can cause phy-

sicians to forget the role of the "traditional," clinically useful techniques involving chromosome or enzyme analysis.

The second source of confusion is that both the popular press and the peer-reviewed medical literature tend to focus on "headline" discoveries ("New gene leads to hope for cancer cure") or on scaremongering ("Job-discrimination fears plague genetic testing"). The hype that surrounds gene discoveries is due in part to "the Gutenberg gap"—the lag time between discovery and publication of the data. Thus, a scientist writing about a gene discovery is given license to imply a direct, immediate application to patient care because it is possible that by the time the manuscript wends its way through peer review, editorial revisions, galley proofs, publication, and distribution, this discovery may indeed be clinically applicable. It is no wonder that clinicians, barraged with all of this information and opinion, struggle to find the best evidence available to formulate evolving standards of care. Fed a rich daily diet of gene discovery and ethical dilemmas, they lack basic information on the uses of genetic testing and the tenets of genetic counseling that will allow them to incorporate these concepts into their practice.

As consultants to the Genetics in Primary Care Project, a federally funded endeavor to help faculty members in primary care residency programs become familiar with medical genetics, we believe there are 3 ways practitioners can better care for their patients with inherited diseases. The first is improved case recognition of patients presenting with symptoms and signs that are manifestations of common genetic diseases. The second is greater comfort when responding to patients' requests for information about inherited diseases. The third is fuller awareness of routine genetic screening measures, such as newborn screening for inborn errors of metabolism and congenital hearing loss and preconception screening in populations at high risk. We hope that *Gene Scene* will provide the framework for applying these genetic principles to primary care.